

Pathways

The Newsletter for Glycoprotein Storage Diseases

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website www.ismrd.org

Our Mission and Vision

ISMRD is the leading advocate for families world-wide affected by Glycoprotein Storage Diseases.

Through partnerships built with medicine, science and industry we seek to detect and cure these diseases, and to provide a global network of support and information.

We seek a future in which children with Glycoprotein storage disease can be detected early, treated effectively and go on and live long healthy and productive lives.

ISMRD supports the following disorders

Alpha Mannosidosis, Aspartylglucosaminuria, Beta Mannosidosis, Fucosidosis, Galactosialidosis, Mucopolipidosis II alpha/beta(I-Cell Disease), Mucopolipidosis III alpha/beta (Pseudo-Hurler Polydystrophy), Mucopolipidosis III Gamma, Schindler Diseases and Sialidosis

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<https://www.facebook.com/groups/82945687520/>

<https://twitter.com/ISMRD>

Donations ISMRD is a 501(c) charitable organisation based in the United States serving a global constituency. We provide our services, which include our newsletter, website, outreach activities and support of research, without requesting monthly dues or any other financial restrictions. We gratefully accept donations that will enable us to continue toward our goal of a future free of the tragic consequences of Glycoprotein Storage Diseases.

Donations can be made via our website using



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From the President's Desk

*By Mark Stark
President, ISMRD*



Welcome to the ISMRD October newsletter! As you will see in the newsletter, we are all very excited to announce that the Fourth International Conference on Glycoproteinoses is scheduled for July 23rd to 26th, 2015 in St. Louis, Missouri. The conference will further our mission at ISMRD to help all those affected by rare diseases by building partnerships with medicine, science and industry in methods to detect and cure these diseases, and to provide a global network of support and information. Our board is working very hard to make sure it is informative and fun for all participants.

Our scientific board has done a great job of bringing together experts from around the world, and all of our board members and volunteers are working to make this the best conference yet for both researchers and families. The hard work has started. We are very pleased with the commitment to support the conference from both corporations and individuals, but more is needed to ensure young researchers and less fortunate families can attend the conference. We very much need your help, and this newsletter includes some easy ways that you can help contribute to the conference. If you would like to volunteer your time, please contact me or one of our other board members. ISMRD is an all-volunteer organization with a very hard-working board, but we do need our members' help.

I would like to welcome several new families to ISMRD that are listed in the newsletter. I would also like to wish speedy recovery to some of our members who have recently been in the hospital. I hope you will join me in offering condolences to the family of Hannah Voltz.

Also included in this newsletter are links to several articles about newborn screening for rare diseases and drugs for rare diseases. As these articles show, there is a tremendous opportunity to use advanced technology to detect, treat, and eventually cure these diseases. Also, I am very happy to note that one of our members, Dee Crompton, has written and had published a book about being the parent of a child with rare diseases. As always, this newsletter contains helpful links to resources, and a schedule of upcoming events.

I would like to again thank everyone who has already contributed this year to ISMRD; both financially and with your time. I hope to meet all of you in St. Louis!

Thank you

**Mark Stark
President
ISMRD**



ISMRD's **MEET US IN ST. LOUIS**

JULY 23 - 26TH 2015 VENUE: HILTON AT THE BALLPARK, ST. LOUIS, MISSOURI, USA

FOR THE 4TH **INTERNATIONAL CONFERENCE ON GLYCOPROTEINOSES**

Washington University in St. Louis
SCHOOL OF MEDICINE

Crossing oceans for a cure

Can you feel the excitement building for this conference?

The Board of Directors can. Families as far away as Brazil have been asking about the conference.

This is a meeting not to be missed. Dr Kornfeld and his team have built a very strong scientific program and the board are developing an exciting and interactive family program which we think will cater to everyone's needs.

The Children's program is going to be one the children/adults will never forget. Of course we can't give all our surprises away.

Many of you have been asking about Registration costs, Scholarships and when registration will open etc. So here we go.

Registrations will open 1st January 2015.

Registration Costs: Early bird Family per person \$125.00USD. **Late Registration Family** per person \$150.00 USD. Unaffected children per child \$50.00USD, Affected children/Adults – No charge.

Accommodation bookings: We are still working with the hotel on this one. Conference accommodation bookings will be able to be done on-line and will open 1st July to coincide with the opening of registration.

Scholarships: This too will be available from 1st January 2015 and will be an on-line application to the ISMRD Board of Directors. Once we have the requests in, we will determine the need and the amount that we can assist.

For families wanting to get a head start in raising funds to help meet some of the costs of attending the conference we have provided a fundraiser below. This fundraiser has a mind of its own and is taking off, don't miss out on this incredible opportunity to raise funds to help cover all or some of your accommodation costs.

If you want more information please contact us at info@ismrd.org

Jenny Noble
Vice President/Administration

Raise money for your conference accommodation costs

For all our families who are thinking of coming to the conference in 2015, here is a suggestion for helping to meet your accommodation costs



We are offering this beautiful bracelet as a fundraiser to help you raise funds to pay for your accommodation. The bracelet is made up with a teal ribbon, and silver-tone and imitation pearl beads that surround three silver-tone charms that say "Hope", "Strength", and "Courage". It measures 8" and stretches to fit most wrists.

The bracelets sells for USD\$20. For every bracelet you sell, ISMRD will put aside USD\$10 in your name to cover some or all of your accommodation.

For example if you sell 30 bracelets, you will have USD\$300 towards your accommodation.

If you would like to purchase some bracelets to sell to your friends and family, please send an [e-mail to info@ismrd.org](mailto:info@ismrd.org). We can send them to you in bulk for you to distribute, or we can send them to the individual buyers, if you provide us with their addresses. Payment can be made via PayPal on our website.

If you are not attending the conference, you can sell bracelets and nominate a family that is attending to receive the \$10 towards their accommodation costs.

The bracelets are very popular and are selling well



Don't forget the ISMRD gofundme page, which has so far raised US\$2470. Every little bit helps. If you would like to donate, go to <http://www.gofundme.com/5rpihw>



ISMRD Calendar 2015

Submit Your Photos Now



ISMRD's 2014 calendar was a big success. It showed photos of 60 ISMRD members from around the world. We sold over 500 calendars to seven countries. As a result of that resounding outcome, we have decided to produce a 2015 calendar.

If you would like your child or affected person to appear in the 2015 calendar, please send us one of your favorite photos of them. Photos should be submitted electronically in the highest resolution possible, and include your child's name, and birthday in MM/DD/YY format. We will put each person's photo under the month of their birthday. Email the photo and details to vivianrichmond@thespecialistworks.com. We would like to thank ISMRD member Martin Woolley (father to Saffy who has Alpha Mannosidosis) for his offer to coordinate the calendar this year.

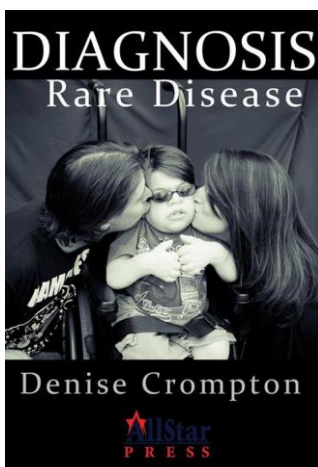
If you have any questions, please contact Susan Kester at kickifer1@aol.com.



ISMRD Member Dee Crompton's book about Mucopolysaccharidosis, *Diagnosis: Rare Disease* has been published

Here is what Dee has to say:

In January 2011, I reached out to ask other ML families if they wanted to participate in my new book, *Diagnosis: Rare Disease*. When twelve mothers responded, I knew that this was a project worthy of my time. They all answered my many questions as I put together a manuscript to tell the stories about what it is like for their families to live with a rare disease.



Thanks to the mothers of Zachie Haggett, Sergio Cardenas, Spencer Gates, Anna James, Joey Nagy, Andre Andrews, Jennifer Klein, Callie Nagle, Sammy and Huddy Anthony, Sarah and Hayden Noble, Allison Dennis and Autumn Tobey, I was able to put together a comprehensive view of family life when at least one member has a rare disease.

Diagnosis: Rare Disease has a twofold purpose. One is to help increase awareness by documenting the many ways in which these families have been affected by ML, and the second is to raise funds to be put toward research. All Star Press released the ebook version of the book on September 15, 2014, and the paper version will most likely be available the following month.

There is also a Facebook page specifically for this subject. All who are interested in rare diseases are welcome here: <https://www.facebook.com/diagnosis.raredisease>



Competitions

Submit Your Artwork! Expression of Hope Now Open for Submissions



The [Expression of Hope III](#) program is now open for submissions. Expression of Hope is a global program of awareness and inspiration featuring works of art by the Lysosomal Storage Disorder (LSD) community. Artists of all ages and artistic ability -- including patients, caregivers, family members, friends, and healthcare providers -- who have been affected by an LSD can submit an original work of art that reflects their own individual story.

This unique program encourages anyone affected by a lysosomal storage disorder to create and submit a work of art that expresses their experiences of living with an LSD. Through the Expression of Hope program, Genzyme furthers its long-standing history of collaboration with patient advocacy organizations around the world to help raise awareness and build a stronger global network of support for people affected by LSDs.

Entries close on 5 December 2014. To read more, go to

http://expressionofhope.com/?utm_source=Genzyme+Rare+Community&utm_campaign=9bca310471-RSS_EMAIL_CAMPAIGN&utm_medium=email&utm_term=0_671f196937-9bca310471-85568409



"Living with a Rare Disease" Photo Contest

Anyone with an interest in rare diseases can participate in the EURORDIS Photo Contest 2014. The Photo Contest gives participants a chance "to communicate visually the many diverse facts of living with a rare disease."



EURORDIS
Rare Diseases Europe

If you are not a photographer, you can participate by voting for your favorite photographs.

An Apple iPad Air tablet will be awarded to each winner of the following three categories:

- **EURORDIS Favorite:** Chosen by EURORDIS staff
- **Public Vote:** The photograph that receives the most votes
- **Expert's Choice:** Selected by Rick Guidotti, professional fashion photographer.

The competition closes in December.

To read more, go to <http://orphandrugonaut.wordpress.com/2014/09/18/eurordis-living-with-a-rare-disease-photo-contest/>



What's happening around the world?

Genetic Alliance Wins Multi-Million Dollar Newborn Screening Award

Renewed Commitment to Run Nation's Newborn Screening Clearinghouse

Genetic Alliance has been awarded a four-year cooperative agreement to maintain and build upon the USA's Newborn Screening Clearinghouse. This cooperative agreement builds upon the original award for the Newborn Screening Clearinghouse, obtained by Genetic Alliance in 2009.

This next iteration of the Clearinghouse will focus on both raising awareness for newborn screening and best practices for educational initiatives.

To read more, go to <http://us5.campaign-archive1.com/?u=a9e55c5fcbe8bce63e2c83eb9&id=e3da57df89>



How Much Money is Spent on Orphan Drugs?

If you think there is no money in orphan drug sales, see the table below from "Biotech Report: Ultra Man", from the August 2014 issue of Medical Marketing & Media.

Top 10 Companies By Orphan Drug Sales (2013)

Rank	Company	Global Sales(\$ Billions) *
1	Novartis	\$ 11.4
2	Roche	\$ 9.5
3	Celgene	\$ 5.6
4	Pfizer	\$ 5.2
5	Teva	\$ 5.1
6	Bayer	\$ 4.1
7	Sanofi	\$ 3.2
8	Merck KGaA	\$ 3.0
9	Biogen Idec	\$ 3.0
10	Baxter International	\$ 2.9

* Sales, EvaluatePharma (per MM&M article chart)

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To read more, go to <http://www.mmm-online.com/biotech-report-ultra-man/article/362218/>



Orphan Drugs: Is There A Relationship Between Disease Prevalence And Price?

Much discussion has recently centered on the pricing of orphan drugs. Why are the prices of orphan drugs and of other new medications (especially for oncology indications) so high? Is there a relationship between the price of an orphan drug and the prevalence of the rare disease it treats?

A report, from Thomson Reuters, [Lysosomal Storage Disease: An example of a lucrative ultra-orphan market](#), by Ulrike Jahnke, reviews the LSD market and the advantages of ultra-orphan therapies. One interesting topic addressed in the report is the question of whether or not there is a relationship between the prevalence of a rare disease and the corresponding price of the disease's approved drug.

The following chart was created from data in the report:

Correlation Between Prevalence And Price

Drug Name and Type of Therapy	Indication	Sponsor Company	Approximate this number of Patients	Price per Patient per Year
Naglazyme ERT* Injection	Maroteaux-Lamy Syndrome (MPS VI)	BioMarin	1,100	\$375,000
Elaprase ERT Injection	Hunter Syndrome (MPS II)	Shire	2,000	\$375,000
Vimizim ERT Injection	Morquio A (MPS IV A)	BioMarin	3,000	\$380,000
Aldurazyme ERT Injection	Hurler (MPS I)	Genzyme (Sanofi)/ BioMarin	3,000 – 4,000	\$200,000
Fabrazyme ERT Injection	Fabry Disease	Genzyme (Sanofi)	5,000 – 10,000	\$200,000
Replagal ERT Injection	Fabry Disease	Shire	5,000 – 10,000	\$150,000 – \$200,000
Myozyme ERT Injection	Pompe Disease	Genzyme (Sanofi)	< 10,000	\$300,000
Cerezyme ERT Injection	Gaucher Disease	Genzyme (Sanofi)	< 10,000	\$200,000
Vpriv ERT Injection	Gaucher Disease	Shire	< 10,000	\$170,000
Ellelyso ERT Injection	Gaucher Disease	Pfizer/ Protalix Biotherapeutics	< 10,000	\$150,000
Cerdelga ** Oral Pill	Gaucher Disease	Genzyme (Sanofi)	<10,000	\$310,250

* ERT = Enzyme Replacement Therapy. ** Cerdelga data not from report. Copyright © 2012-2014, [Orphan Drugonaut Blog](#). All rights reserved.

Mexico's Top 7 Most Common Rare Diseases



It's estimated that about 7 million Mexicans have a rare disease and that only 500 of them have been diagnosed. This is due to several factors such as lack of knowledge about the disease, misdiagnosis, difficulty in accessing health care, and availability of treatments.

The following are the 7 most common rare diseases in Mexico:

- Myelofibrosis:
- Cushing Disease
- Tuberous Sclerosis
- Acromegaly
- Fabry Disease
- Gaucher's Disease
- Mucopolysaccharidosis Type 1

Read more: <http://www.publimetro.com.mx/noticias/mexicanos-sin-atencion-en-enfermedades-raras/mnbB!c243qHYCRHYLc/> (in Spanish)

How to make a Penguin

(Thank you, Rhonda Skipper, for sharing this with us)





Carer Resources

Health-related SmartPhone Apps

[PatientView](#), an organization founded in 2000, with the “.. purpose of understanding the most significant factor to affect healthcare in the 21st century – the patient movement ...”, publishes a free downloadable report entitled, [European Directory of Health Apps 2012-2013](#). The Directory contains detailed facts and information on smartphone health apps that help patients manage their medical conditions. These are healthcare apps recommended by healthcare communities from all over the world, and can be viewed in 47 languages.

For more information, go to <http://orphandrugonaut.wordpress.com/2014/07/31/patient-advocacy-reaching-out-through-smartphone-apps/>

New Online Resource for Patient Advocates Working to Ensure Patient Voice is Heard

Patient advocates who want to ensure the patient experience is incorporated into the complex world of drug evaluation and medicines development now have a new resource to assist them. The [eMEET \(Medicine Evaluation Educational Training\)](#) is designed to help patient advocates navigate the complex world of medicines development, evaluation and assessment.

To read more, go to

http://www.meetforpatients.com/?utm_source=Genzyme+Rare+Community&utm_campaign=44a35ad974-RSS_EMAIL_CAMPAIGN&utm_medium=email&utm_term=0_671f196937-44a35ad974-85568409

Rare Disease Patient Voices in the Clinical Trials process

To read about the effect a rare disease patient advocate can have on the Clinical Trials process, read Steve Smith's story at: <http://www.forbes.com/sites/medidata/2014/09/25/rare-disease-patient-voices-bring-change-to-the-clinical-trials-process/>

The Firefly

Go to <https://www.facebook.com/video.php?v=355842444455037&set=vb.100000878532027&type=2&theater> to see a video of the Firefly, which converts a standard wheelchair into a motorized scooter.

Free personalized songs for children and teens







The Songs of Love Foundation is a nonprofit organization dedicated to providing personalized songs for children and teens currently facing tough medical, physical or emotional challenges, free of charge.

Go to <http://www.songsoflove.org/> (Thank you Danielle Z for this tip)



ISMRD warmly welcomes the following
people to our family



-  Cheryl Closs and her daughter Cheryl Ann Dockery (nickname Baby Bella, aged 11 years) who has Fucosidosis. They live in Florida, USA. Also her grandmother Loreen Kemp and cousin Mary Masterton who have joined our family.
-  Bobbie Gross, who has two children with Alpha Mannosidosis, a 14 year old girl and a 19 year old boy. They live in Virginia, USA.
-  Anna Palomo, who has a 23 year old daughter with Alpha Mannosidosis and a son who had Alpha Mannosidosis who passed away when he was 18. They live in Texas, USA
-  Diane Roncone
-  Laila Shannon and her son Sari, who is 24 years old and has Alpha Mannosidosis
-  Julia Taravella who has two boys with Aspartylglucosaminuria.



With Deepest Sympathy



May the stars carry your sadness away,
May the flowers fill your heart with beauty,
May hope forever wipe away your tears,
And, above all, may silence make you strong

Sadly we mourn the loss of

Hannah Voltz

who passed away on 28 August 2014, age 10.
Hannah had Mucopolidosis II









If you know of anyone who has recently been ill or had surgery or is about to have surgery, please tell us at info@ismrd.org



Some of our Penguin children and young adults have recently been in hospital, had surgery or are awaiting surgery

Your Penguin family are thinking of you and praying for a good outcome

-  Jenny Klein, ML III
-  Mashudu Hlatshwayo, ML II
-  Callie Nagle, ML III, hip reconstruction surgery
-  Sergio Cardona-Cardenas, ML II/III



Create something at Live!uvCreate.com

Thank you to Sharon Meador for this

ISMRD'S Sunshine Care Committee



ISMRD has a group of parent volunteers called the **"Sunshine Committee"**. Our purpose is to coordinate support for families in need. The type of support varies on the circumstance -- from birthday and weddings, an illness or death in the family, or a family experiencing surgery or a medical crisis. In any case, we provide a little "sunshine" for the family by providing flowers, encouraging messages via email, cards or a phone call -- whatever we think the family would find most helpful. In order to help others, our group relies on the support of all families because, in essence, we are all part of the ISMRD "Sunshine Committee".

If you are in need of assistance or know someone in our Penguin community who is in need, **please contact Susan Kester**. She will coordinate with the appropriate parties to determine how we can best help.



ISMRD gratefully acknowledges the following people for their very generous donations.

Without this kind of support we would not be able to carry out our mission and vision for ISMRD.

- | | |
|---|--|
|  Tish Adkins |  Kris McKenna, in memory of Kendall & Kylie Moran |
|  Ann |  Anne Marie Mead |
|  Ann Marie Arquilla |  Parker Meador |
|  Larry & Barbara Arquilla |  Anita Muonio |
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|  Michelle McGee |  Martin Woolley |